

Disease

Alkaptonuria^{1, 2, 3}

Gene

HGD

Inheritance	AR
Current Treatments	Nitisinone, diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urinary homogentisic acid ⁴
Disease Frequency (per 100,000)	0.25 ⁴
Genes Included in Disease Frequency	HGD
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Alkaptonuria&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/203500>

² <https://pubmed.ncbi.nlm.nih.gov/12501223/>

³ <https://pubmed.ncbi.nlm.nih.gov/34344451/>

⁴ <https://www.ncbi.nlm.nih.gov/books/NBK1454/>